遗传　2003 25 (5): 538-542　ISSN: 0379-4172　CN: 11-5450/R

阿尔茨海默病中β淀粉样前体蛋白基因外显子9、10的mRNA表达及突变研究 Expression Variation and Mutation Analysis of Exon 9 and 10 in Amyloid Precursor Protein Gene in Alzheimer's Disease

江三多1,汤国梅1,张明园1,任大明2,钱伊萍1,张野1,林嗣萃1,李飞1,汪栋祥1 JIANG San-Duo1,TANG Guo-Mei1,ZHANG Ming-Yuan1,REN Da-Ming2,QIAN Yi-Ping1,ZHANG Ye1,LIN Si-Cui1,LI Fei1,WANG Dong-Xiang1

1.上海市精神卫生中心,上海 200030; 2 复旦大学遗传研究所,上海 200433 1.Shanghai Mental Health Center,Shanghai 200030,China; 2.Institute of Genetics,Fudan University,Shanghai 200433,China

收稿日期　修回日期　网络版发布日期　接受日期

摘要　于上海地区汉人群中，探讨阿尔茨海默病(AD)患者外周血中β淀粉样前体蛋白基因外显子9、10（APP9~10）的mRNA表达水平，并作cDNA的点突变检测。采用半定量竞争性反转录聚合酶链反应(semi-quantitive competitive RT-PCR)技术测定APP9~10的mRNA表达水平；以聚合酶链反应（PCR）-限制性片段长度多态(RFLP)方法检测基因组DNA中载脂蛋白E基因(APOE)和早老素1基因(PS1)多态；APP9~10cDNA的点突变检测采用变性梯度凝胶电泳法。结果表明（1）AD患者中 APP9~10的mRNA表达水平显著高于健康老人（P＜0.05）；（2）作为AD的主要风险因子，APOE*ε4等位基因并不影响AD患者中APP9~10的mRNA表达水平,而另一可疑风险因子PS1基因1等位基因的存在却可显著增加AD患者中APP9~10的mRNA表达；（3）AD患者中不存在APP9~10cDNA的点突变。 提示上海汉人群中，AD患者外周血中APP基因外显子9、10的mRNA表达水平显著升高，也许这一生物学改变可作为一种生物学辅助诊断指标应用于AD的临床研究。

关键词：阿尔茨海默病；β淀粉样前体蛋白基因；表达；突变

Abstract:To explore the expression differences of exon 9 and 10 in Amyloid Precursor Protein gene (APP9~10) in Alzheimer′s disease,and detect the probable point mutation appeared in cDNA fragment of APP9~10 in the Shanghai Han people.semi-quantitaive competitive RT-PCR technique was performed to detect the expression of APP9~10 in peripheral lymphocyte,and the Apolipoprotein E gene(ApoE) and Presenilin 1(PS1)gene were genotyped with PCR-RFLP method.We also analyzed the point mutation in APP9~10 cDNA through the denatured gel eletrophoresis.The results are as follows:1.While compared with healthy controls,expression of APP9~10 mRNA was significantly enhanced in Alzheimer disease;2.APOE*ε4 allele,the most common genetic risk factor for AD,did not affect the Expression of APP9~10 mRNA,whereas the APP9~10 mRNA expression might be increased by the allele 1 of PS1 gene,another probable susceptibility gene of AD.3.No point mutation in APP9~10 cDNA was detected.In our samples,the expression of APP9~10 mRNA in AD was significantly different from that of controls,suggesting that the change of peripheral APP9~10 mRNA expression might be another bio-marker used in clinical diagonosis for AD.

关键词　Key words　Alzheimer's disease　amyloid precursor protein gene　expression　mutation

分类号

Abstract

Key words

DOI:

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